INVITAE

INVITAE DIAGNOSTIC TESTING RESULTS

Patient name:	H[redacted] Almoosi	**Sample type:**	Buccal Swab	**Report date:**	12/10/2021
DOB:	[redacted]/2018	**Sample collection date:**	11/19/2021	**Invitae #:**	RQ2902695
Sex assigned at birth:	Female	**Sample accession date:**	11/22/2021	**Clinical team:**	Alexandra Dunn Julie Ziobro
Gender:		**MRN:**	101484343		

Reason for testing

Diagnostic test for a personal history of disease

Test performed

Sequence analysis and deletion/duplication testing of the 302 genes listed in the Genes Analyzed section.

- Invitae Epilepsy Panel

RESULT: POTENTIALLY POSITIVE

Two Pathogenic variants identified in TPP1. TPP1 is associated with autosomal recessive neuronal ceroid lipofuscinosis.

Additional Variant(s) of Uncertain Significance identified.

GENE	VARIANT	ZYGOSITY	VARIANT CLASSIFICATION
TPP1	c.1094G>A (p.Cys365Tyr)	heterozygous	PATHOGENIC
TPP1	c.1558del (p.Arg520Valfs*12)	heterozygous	PATHOGENIC
KCNJ10	c.305C>G (p.Pro102Arg)	heterozygous	Uncertain Significance
KCNMA1	c.36_65del (p.Gly13_Ser22del)	heterozygous	Uncertain Significance
KIF1A	c.1290C>T (Silent)	heterozygous	Uncertain Significance
PLAA	c.1445C>T (p.Ser482Leu)	heterozygous	Uncertain Significance
RELN	c.6554T>A (p.Met2185Lys)	heterozygous	Uncertain Significance

About this test

This diagnostic test evaluates 302 gene(s) for variants (genetic changes) that are associated with genetic disorders. Diagnostic genetic testing, when combined with family history and other medical results, may provide information to clarify individual risk, support a clinical diagnosis, and assist with the development of a personalized treatment and management strategy.

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